

CLAIMS

1. A method for the diagnosis of a single nucleotide polymorphism in a pyruvate dehydrogenase kinase isoenzyme 2 (PDK2) gene in a human, which method comprises
- 5 determining the sequence of the nucleic acid of the human at one or more of positions 288, 1281 and 1357 in the PDK2 gene as defined by the positions in EMBL ACCESSION NO. L42451, and determining the status of the human by reference to polymorphism in the PDK2 gene.
- 10 2. A method for diagnosis according to claim 1 in which the single nucleotide polymorphism is further defined as:
- the single nucleotide polymorphism at position 288 is presence of C and/or T;
- the single nucleotide polymorphism at position 1281 is presence of G and/or A;
- the single nucleotide polymorphism at position 1357 is presence of G and/or C.
- 15 3. A method for diagnosis according to claim 1 or 2 in which the sequence is determined by a method selected from amplification refractory mutation system and restriction fragment length polymorphism.
- 20 4. Use of a method according to any of claims 1 - 3 for predicting the clinical response to a therapeutic compound, or for determining the therapeutic dose of a compound, in the treatment of PDK2- mediated disease.
5. Use of a method according to any of claims 1 - 3 for assessing the predisposition of an
- 25 individual to diseases mediated by PDK2.
6. A nucleic acid comprising any one of the following polymorphisms:
- the nucleic acid of EMBL ACCESSION NO. L42451 with T at position 288 as defined by the position in EMBL ACCESSION NO. L42451;
- 30 and/or

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the nucleic acid of EMBL ACCESSION NO. L42451 with A at position 1281 as defined by the position in EMBL ACCESSION NO. L42451;

and/or

the nucleic acid of EMBL ACCESSION NO. L42451 with C at position 1357 as defined by

5 the position in EMBL ACCESSION NO. L42451;

or a complementary strand thereof or an antisense sequence thereto or a fragment thereof of at least 20 bases comprising at least one polymorphism.

7. An allele-specific primer capable of detecting a PDK2 gene polymorphism at one or
10 more of positions 288, 1281 and 1357 in the PDK2 gene as defined by the position in EMBL ACCESSION NO. L42451.

8. An allele-specific oligonucleotide probe capable of detecting a PDK2 gene
polymorphism at one or more of positions 288, 1281 and 1357 in the PDK2 gene as defined
15 by the positions in EMBL ACCESSION NO. L42451.

9. A diagnostic kit comprising an allele-specific primer as defined in claim 7 or an allele-specific oligonucleotide probe as defined in claim 8.

20 10. A method of treating a human in need of treatment with a PDK2 drug in which the method comprises:

(i) diagnosis of a single nucleotide polymorphism in the PDK2 gene in the human, which diagnosis comprises determining the sequence of the nucleic acid at one or more of positions 288, 1281 and 1357 of the PDK2 gene as defined by the positions in EMBL ACCESSION
25 NO. L42451, and determining the status of the human by reference to polymorphism in the PDK2 gene;

and

(ii) administering an effective amount of a PDK2 drug.

30 11. Use of a PDK2 drug in the preparation of a medicament for treating a PDK2-mediated disease in a human diagnosed as having a single nucleotide polymorphism at one or more of

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positions 288, 1281 and 1357 of the PDK2 gene as defined by the positions in EMBL
ACCESSION NO. L42451.

12. A computer readable medium comprising at least one nucleic acid sequence as defined
5 in claim 6 stored on the medium.

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